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MASTERING COMMUNITY MEDICINE

High-Yield MCQs with Explanations

A Tool for Competency Driven Assessment in Community Medicine

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JAYPEE

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21 CHAPTER

Genetics and Health

Roopali Nath Mathur, Purushottam Giri

SPECIFIC LEARNING OBJECTIVES (SLOs)

1. Understand basic genetic concepts and inheritance.

Learners will be able to explain fundamental genetic terms and patterns of inheritance, enabling them to understand how genetic traits and disorders are transmitted across generations.

2. Identify genetic disorders of public health importance.

Learners will recognize common genetic conditions prevalent in India, particularly hemoglobinopathies, and understand their epidemiological significance.

3. Explain principles of genetic screening and counseling.

Learners will be able to describe various screening strategies and the role of genetic counseling in prevention and reduction of genetic disease burden.

4. Discuss the Human Genome Project and ethical issues.

Learners will understand the objectives, outcomes, and ethical, legal, and social implications of advances in human genetics.

5. Apply genetics in community health and prevention.

Learners will be able to integrate genetic knowledge into public health planning, national programs, and preventive strategies, including relevant legal provisions.

MULTIPLE CHOICE QUESTIONS (MCQs)

1. **A couple who are both carriers of β-thalassemia plan another pregnancy. What is the probability that their next child will be affected?**
   A. 0%
   B. 25%
   C. 50%
   D. 75%

2. **A boy presents with recurrent hemarthrosis and family history in maternal uncle. What is the inheritance pattern?**
   A. Autosomal dominant
   B. Autosomal recessive
   C. X-linked recessive
   D. Multifactorial

3. **During RBSK screening, a newborn is diagnosed with cleft palate. Under which category of the 4Ds does this condition fall?**
   A. Disease
   B. Birth defect
   C. Deficiency
   D. Developmental delay

4. **A tribal screening program identifies several adolescents with sickle cell trait. What is the most appropriate preventive strategy?**
   A. Blood transfusion
   B. Carrier screening and counseling
   C. Chemotherapy
   D. Isolation

5. **A newborn presents with hypotonia and epicanthic folds. Which associated anomaly should be screened first?**
   A. Congenital heart disease
   B. Asthma
   C. Tuberculosis
   D. Renal stones

6. **Random changes in allele frequencies in a small population are called:**
   A. Mutation
   B. Genetic drift
   C. Natural selection
   D. Migration

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7. A girl with short stature and primary amenorrhea has Turner syndrome. What is the karyotype?
A. 45 XO
B. 47 XXY
C. 47 XYY
D. 46 XX

8. Consanguineous marriage increases the risk of:
A. Autosomal dominant disorders
B. Autosomal recessive disorders
C. Infectious diseases
D. Trauma

9. Genetics is defined as the study of:
A. Microorganisms
B. Heredity and variation
C. Nutrition
D. Environment

10. Which supplement helps prevent neural tube defects?
A. Iron
B. Folic acid
C. Calcium
D. Vitamin C

11. Who proposed the double helix model of DNA?
A. Mendel
B. Darwin
C. Watson and crick
D. Lamarck

12. The main objective of community genetics is:
A. Individual treatment
B. Population prevention
C. Laboratory research
D. Surgical management

13. The basic unit of heredity is:
A. Chromosome
B. Gene
C. Protein
D. RNA

14. Which intervention helps reduce sickle cell disease incidence?
A. Vaccination
B. Carrier detection
C. Antibiotics
D. Surgery

15. Coefficient of inbreeding indicates:
A. Mutation rate
B. Probability of identical alleles
C. Migration rate
D. Fertility rate

16. Rashtriya Bal Swasthya Karyakram is implemented under:
A. NACP
B. NHM
C. NTEP
D. NPCDCS

17. Mutation may be caused by:
A. Radiation
B. Chemicals
C. Viruses
D. All of the above

18. The concept of genetic load refers to:
A. Total number of genes
B. Proportion of harmful alleles in population
C. Chromosomal mutation only
D. Burden of counseling

19. Development of disease due to genetic susceptibility and lifestyle factors is called:
A. Mutation
B. Gene–environment interaction
C. Genetic drift
D. Chromosomal disorder

20. Population genetic screening programs in India mainly focus on:
A. Hemoglobinopathies
B. Chromosomal disorders
C. Metabolic disorders
D. Infectious diseases

21. A pedigree chart is mainly used to study:
A. Infection patterns
B. Inheritance pattern
C. Nutritional status
D. Fertility pattern

22. The most common chromosomal abnormality seen in Down syndrome is:
A. Trisomy 21
B. Trisomy 18
C. Monosomy X
D. Trisomy 13

23. The first affected individual identified in a pedigree is called:
A. Carrier
B. Proband
C. Ancestor
D. Index case

24. How many pairs of autosomes are present in humans?
A. 22
B. 23
C. 24
D. 21

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25. According to Hardy–Weinberg law, if disease frequency is 1/10,000, carrier frequency is approximately:
A. 1/50 B. 1/25
C. 1/100 D. 1/200

26. Amniocentesis is usually performed at which gestational age?
A. 6–8 weeks
B. 10–12 weeks
C. 15–18 weeks
D. 30 weeks

27. Premarital genetic screening helps prevent:
A. Infectious diseases
B. Genetic disorders
C. Nutritional diseases
D. Occupational diseases

28. Which Mendelian law states that alleles separate during gamete formation?
A. Law of dominance
B. Law of segregation
C. Law of linkage
D. Law of mutation

29. The father of genetics is:
A. Darwin
B. Mendel
C. Lamarck
D. Pasteur

30. India's National Policy for Rare Diseases mainly focuses on:
A. Infectious disease control
B. Management of rare genetic disorders
C. Sanitation
D. Vector control

31. The most important risk factor for Down syndrome is:
A. Paternal age
B. Maternal age
C. Birth order
D. Socioeconomic status

32. Cri-du-chat syndrome is caused by:
A. Trisomy 21
B. Deletion of chromosome 5p
C. Monosomy X
D. Trisomy 18

33. Hardy–Weinberg equilibrium explains:
A. Mutation
B. Stable allele frequency
C. Migration
D. Fertility

34. Klinefelter syndrome has which karyotype?
A. 45 XO
B. 47 XXY
C. 47 XYY
D. 46 XY

35. First trimester screening for chromosomal abnormality includes:
A. Triple test
B. Double marker test
C. Hb electrophoresis
D. Blood culture

36. A mutation involving change of one nucleotide is called:
A. Chromosomal mutation
B. Point mutation
C. Polygenic mutation
D. Structural mutation

37. Which screening criteria include that disease must be important health problem?
A. Mendel criteria
B. Wilson and Jungner criteria
C. Darwin criteria
D. Koch criteria

38. Edwards syndrome results from:
A. Trisomy 21
B. Trisomy 18
C. Trisomy 13
D. Monosomy X

39. Increased disease frequency due to small founding population is called:
A. Genetic drift
B. Founder effect
C. Mutation
D. Selection

40. Patau syndrome is caused by:
A. Trisomy 13
B. Trisomy 18
C. Trisomy 21
D. XO

41. A mutation causing amino acid change is called:
A. Silent mutation
B. Missense mutation
C. Nonsense mutation
D. Frameshift mutation

42. Higher genetic disease prevalence is commonly seen in:
A. Urban population
B. Migrant population
C. Isolated population
D. Mixed population

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43. Prenatal diagnosis of chromosomal disorders is confirmed by:
A. Ultrasound
B. Amniocentesis
C. Blood sugar
D. X-ray

44. Genetic information from DNA to ribosome is carried by:
A. tRNA
B. mRNA
C. rRNA
D. DNA polymerase

45. Which Act regulates prenatal diagnostic techniques in India?
A. MTP Act
B. PCPNDT Act
C. Epidemic Act
D. Food Safety Act

46. Presence of identical alleles is called:
A. Heterozygosity
B. Homozygosity
C. Mutation
D. Linkage

47. RBSK screening covers children up to:
A. 5 years
B. 10 years
C. 18 years
D. 25 years

48. Gene therapy involves:
A. Drug therapy
B. Replacement of defective gene
C. Nutrition therapy
D. Radiation therapy

49. Genetic material in human cells is mainly located in:
A. Mitochondria
B. Nucleus
C. Golgi body
D. Lysosome

50. Diseases like diabetes and hypertension show:
A. Autosomal dominant inheritance
B. Autosomal recessive inheritance
C. Multifactorial inheritance
D. X-linked inheritance

51. A child with Duchenne muscular dystrophy shows which inheritance pattern?
A. Autosomal dominant
B. Autosomal recessive
C. X-linked recessive
D. Multifactorial

52. Which nitrogenous base is present in DNA but not in RNA?
A. Adenine
B. Guanine
C. Thymine
D. Cytosine

53. Increased nuchal translucency is associated with:
A. Down syndrome
B. Malaria
C. Tuberculosis
D. Asthma

54. Observable characteristics of an individual are called:
A. Genotype
B. Phenotype
C. Mutation
D. Allele

55. Carrier detection of β-thalassemia is done by:
A. Blood culture
B. Hemoglobin electrophoresis
C. Blood urea
D. Urine test

56. Gametes are produced by:
A. Mitosis
B. Meiosis
C. Budding
D. Binary fission

57. Mendel's law explaining independent transmission of traits is:
A. Law of dominance
B. Law of segregation
C. Law of independent assortment
D. Law of mutation

58. Structural RNA of ribosome is:
A. mRNA
B. tRNA
C. rRNA
D. siRNA

59. Hardy–Weinberg equilibrium requires:
A. Small population
B. Random mating
C. Mutation
D. Migration

60. Early detection of genetic disorders in children is done under:
A. NTEP
B. RBSK
C. NACP
D. NPCB

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61. Mitochondrial disorders show:
A. Paternal inheritance
B. Maternal inheritance
C. Autosomal inheritance
D. Random inheritance

62. Alternative forms of a gene are called:
A. Phenotype
B. Alleles
C. Mutation
D. Chromosomes

63. A couple with history of three recurrent spontaneous abortions is advised chromosomal analysis. Which investigation is most appropriate to detect a possible genetic cause?
A. ELISA
B. PCR
C. Karyotyping
D. Culture

64. Triple marker test screens for:
A. Tuberculosis
B. Down syndrome
C. Malaria
D. Typhoid

65. Genes are composed of:
A. Protein
B. DNA
C. Lipids
D. Carbohydrates

66. Interaction of genes with environment is called:
A. Mutation
B. Gene expression
C. Gene–environment interaction
D. Drift

67. Mendel conducted experiments on:
A. Wheat
B. Pea plant
C. Rice
D. Maize

68. Carrier detection in families is done using:
A. Pedigree analysis
B. ECG
C. CT scan
D. X-ray

69. Genetic constitution of an individual is:
A. Phenotype
B. Genotype
C. Mutation
D. Allele

70. Newborn screening is commonly done by:
A. Heel prick test
B. ECG
C. MRI
D. CT scan

71. DNA replication is carried out by:
A. RNA polymerase
B. DNA polymerase
C. Ligase
D. Helicase

72. Example of multifactorial disease:
A. Hemophilia
B. Thalassemia
C. Diabetes
D. Turner syndrome

73. Main aim of screening programs is:
A. Treatment
B. Early detection
C. Surgery
D. Rehabilitation

74. Interaction between two genes affecting expression is:
A. Penetrance
B. Expressivity
C. Epistasis
D. Mutation

75. Male sex is determined by:
A. X chromosome
B. Y chromosome
C. Autosome
D. RNA

76. Screening entire population is called:
A. Selective screening
B. Mass screening
C. Diagnostic testing
D. Case finding

77. Formation of RNA from DNA is:
A. Translation
B. Replication
C. Transcription
D. Mutation

78. Variation in severity of genetic disease is called:
A. Mutation
B. Variable expressivity
C. Selection
D. Drift

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79. Only maternal transmission occurs in:
A. Hemophilia
B. Mitochondrial disorders
C. Down syndrome
D. Turner syndrome

80. A 2-year-old child is brought to an RBSK screening clinic with delayed speech and poor social interaction. Under which category of the 4Ds will this child be classified?
A. Deficiency
B. Developmental delay
C. Disease
D. Birth defect

81. A small isolated island population shows unusually high prevalence of a rare metabolic disorder because the population descended from a few ancestors. What is this phenomenon called?
A. Mutation
B. Founder effect
C. Migration
D. Selection

82. Monosomy X results in:
A. Down syndrome
B. Turner syndrome
C. Klinefelter syndrome
D. Edwards syndrome

83. Proportion of genotype expressing phenotype is:
A. Expressivity
B. Penetrance
C. Mutation
D. Drift

84. A 22-year-old tall male presents with infertility, sparse facial hair and gynecomastia. Which investigation confirms the suspected diagnosis of Klinefelter syndrome?
A. Blood sugar
B. Karyotyping
C. Urine test
D. ECG

85. Heritable change without DNA sequence change is:
A. Mutation
B. Epigenetics
C. Drift
D. Selection

86. X-linked recessive disease example:
A. Hemophilia
B. Diabetes
C. Hypertension
D. Asthma

87. A pregnant woman has a high-risk result on triple marker screening. Which diagnostic test should be done to confirm chromosomal abnormality?
A. Ultrasound
B. Amniocentesis
C. Blood pressure
D. CT scan

88. A district genetic screening program identifies carriers of β-thalassemia. What is the major public health benefit of identifying carriers?
A. Treatment
B. Prevention of affected births
C. Surgery
D. Vaccination

89. RNA carrying amino acids is:
A. mRNA
B. rRNA
C. tRNA
D. miRNA

90. Main aim of newborn screening is:
A. Treatment
B. Early detection
C. Surgery
D. Rehabilitation

91. After a natural disaster drastically reduces a village population, the gene frequencies change randomly in subsequent generations. What is this phenomenon called?
A. Selection
B. Bottleneck effect
C. Mutation
D. Migration

92. Mutation of β-globin gene causes:
A. Hemophilia
B. Sickle cell disease
C. Turner syndrome
D. Klinefelter syndrome

93. Testing relatives of affected individuals is:
A. Mass screening
B. Cascade screening
C. Random screening
D. Diagnostic test

94. Mutation due to insertion or deletion is:
A. Silent mutation
B. Missense mutation
C. Frameshift mutation
D. Point mutation

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95. Short stature with streak gonads occurs in:
A. Turner syndrome
B. Down syndrome
C. Edwards syndrome
D. Patau syndrome

96. Separation of alleles during gamete formation is:
A. Law of dominance
B. Law of segregation
C. Law of mutation
D. Law of linkage

97. Mapping of human genes was done by:
A. Apollo project
B. Human genome project
C. Manhattan project
D. Genome India

98. Study of genes inherited together is:
A. Pedigree analysis
B. Linkage analysis
C. ELISA
D. Staining

99. Female genotype is:
A. XY
B. XX
C. YY
D. XO

100. Most important ethical principle in genetic testing is:
A. Profit
B. Confidentiality
C. Publicity
D. Advertising

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ANSWERS WITH EXPLANATION AND REFERENCES

1. Ans: B. 25%

β-thalassemia is an autosomal recessive disorder caused by reduced or absent synthesis of β-globin chains. When both parents are heterozygous carriers, Mendelian inheritance predicts that each pregnancy has a 25% risk of producing an affected child, 50% risk of a carrier child, and 25% chance of a normal child. This probability remains constant for every pregnancy. Carrier screening is an important preventive strategy in India due to high prevalence of hemoglobinopathies. Genetic counseling helps couples understand reproductive risks and options like prenatal diagnosis. Such preventive strategies reduce the long-term public health burden.

[Ref: Park K. Park's Textbook of Preventive and Social Medicine. 28th ed. Jabalpur: Banarsidas Bhanot Publishers; 2025. pp. 612-618.]

2. Ans: C. X-linked recessive

Hemophilia is an inherited bleeding disorder caused by deficiency of clotting factors VIII (Hemophilia A) or IX (Hemophilia B). These genes are located on the X chromosome and follow X-linked recessive inheritance. Males are affected because they possess only one X chromosome, while females usually remain carriers. A characteristic pedigree feature is absence of male-to-male transmission. Clinical manifestations include hemarthrosis and prolonged bleeding after trauma. Early diagnosis helps in factor replacement therapy and genetic counseling.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson and Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 198-205.]

3. Ans: B. Birth defect

RBSK is a child health screening program under the National Health Mission that focuses on early identification of four groups of conditions known as the 4Ds. These include defects at birth, diseases, deficiencies and developmental delays. Cleft palate is a congenital structural anomaly present since birth and therefore classified as a birth defect. Early surgical correction improves feeding, speech and cosmetic outcomes. Early detection reduces long-term disability. RBSK ensures referral and treatment through designated health facilities.

[Ref: Ministry of Health and Family Welfare. Rashtriya Bal Swasthya Karyakram: Operational Guidelines. New Delhi: Government of India; 2013. pp. 14-22.]

4. Ans: B. Carrier screening and counseling

Sickle cell disease is an autosomal recessive disorder caused by a point mutation in the β-globin gene. Individuals with sickle cell trait are usually asymptomatic but can transmit the disease to their offspring. Population carrier screening helps identify at-risk individuals. Genetic counseling allows informed marital and reproductive decisions. Prevention through screening is more effective than treatment. Such programs are particularly important in high-prevalence tribal populations in India.

[Ref: Indian Council of Medical Research. Guidelines for Prevention and Control of Hemoglobinopathies in India. New Delhi: Indian Council of Medical Research; 2018. pp. 20-32.]

5. Ans: A. Congenital heart disease

Down syndrome is caused by trisomy 21 resulting from meiotic nondisjunction. Approximately 40–50% of affected children have congenital heart disease, especially atrioventricular septal defects. Early cardiac evaluation is essential for management. Intellectual disability and thyroid dysfunction are also commonly associated. Early intervention improves survival and quality of life. Prenatal screening programs help early detection and counseling.

[Ref: Jameson JL, Fauci AS, Kasper DL, Hauser SL, Longo DL, Loscalzo J. Harrison's Principles of Internal Medicine. 21st ed. New York: McGraw-Hill Education; 2022. pp. 3115-3128.]

6. Ans: B. Genetic drift

Genetic drift refers to random changes in allele frequencies that occur particularly in small populations. These changes are independent of selective advantage and occur due to chance events. Founder effect and population bottleneck are common examples. Genetic drift may increase prevalence of certain rare genetic disorders. This concept is important in isolated communities. Public health genetic screening programs consider such population characteristics.

[Ref: Park K. Park's Textbook of Preventive and Social Medicine. 28th ed. Jabalpur: Banarsidas Bhanot Publishers; 2025. pp. 618-620.]

7. Ans: A. 45 XO

Turner syndrome is caused by complete or partial monosomy of the X chromosome. Patients commonly present with short stature, gonadal dysgenesis and infertility. Diagnosis is confirmed through karyotyping. Growth hormone therapy may improve final height. Estrogen therapy helps development of secondary sexual characteristics. Early diagnosis helps manage associated cardiovascular and endocrine abnormalities.

[Ref: Jameson JL, Fauci AS, Kasper DL, Hauser SL, Longo DL, Loscalzo J. Harrison's Principles of Internal Medicine. 21st ed. New York: McGraw-Hill Education; 2022. pp. 3128-3132.]

8. Ans: B. Autosomal recessive

Consanguinity increases the probability of inheriting identical alleles from a common ancestor. This increases homozygosity in offspring. As a result, autosomal recessive

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disorders become more common. Examples include thalassemia and metabolic disorders. Community awareness about genetic risks is important. Genetic counseling helps reduce disease occurrence.

[Ref: Bittles AH. Consanguinity and its Relevance to Clinical Genetics. Clin Genet. 2001;60(2):89-98.]

9. Ans: B. Heredity and variation

Genetics is the branch of biology concerned with inheritance of traits from parents to offspring. It also explains variation between individuals. Modern genetics includes molecular genetics, cytogenetics and population genetics. It forms the scientific basis for genetic counseling. Understanding genetics helps prevention of inherited disorders. It is an important component of community medicine.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 3-12.]

10. Ans: B. Folic acid supplementation

Neural tube defects occur due to failure of neural tube closure during early embryogenesis. Folic acid supplementation before conception significantly reduces this risk. WHO recommends supplementation during reproductive age. It is included in routine antenatal care programs. Health education improves compliance among women. This intervention represents an important primary prevention strategy.

[Ref: World Health Organization. Guideline: Daily Iron and Folic Acid Supplementation in Pregnant Women. Geneva: World Health Organization; 2012.]

11. Ans: C. Watson and Crick

James Watson and Francis Crick described the double helix structure of DNA in 1953. DNA consists of two antiparallel strands held together by complementary base pairing. This discovery explained mechanisms of genetic replication and transmission of hereditary information. It laid the foundation of molecular biology and modern genetics. Understanding DNA structure helps explain mutations and genetic diseases. Modern genetic diagnostics are based on these principles.

[Ref: Watson JD, Crick FHC. Molecular Structure of Nucleic Acids: A Structure for Deoxyribose Nucleic Acid. Nature. 1953;171(4356): 737-738.]

12. Ans: B. Population prevention

Community genetics focuses on prevention and control of genetic disorders at the population level. It emphasizes screening, early detection and genetic counseling. The aim is to reduce incidence of inherited diseases. It differs from clinical genetics which focuses on individual management. Public health programs integrate genetics into preventive services. This approach improves long-term population health outcomes.

[Ref: World Health Organization. Community Genetics Services: Report of a WHO Consultation. Geneva: World Health Organization; 2010.]

13. Ans: B. Gene

A gene is a segment of DNA that contains information required to synthesize proteins or functional RNA. Genes determine inherited characteristics. Mutations in genes can result in genetic disorders. Genes are located on chromosomes within the nucleus. Advances in molecular genetics allow identification of disease-causing genes. Genetic testing is based on analysis of gene structure and function.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 5-15.]

14. Ans: B. Carrier detection

Sickle cell disease follows autosomal recessive inheritance. Carrier detection helps identify individuals who can transmit the disease. Genetic counseling allows informed marital and reproductive decisions. Population screening programs help reduce incidence of affected births. Early diagnosis improves management outcomes. This is a key preventive strategy in endemic regions.

[Ref: Indian Council of Medical Research. Guidelines for Prevention and Control of Hemoglobinopathies in India. New Delhi: Indian Council of Medical Research; 2018. pp. 20-32.]

15. Ans: B. Probability of identical alleles

The coefficient of inbreeding measures the probability that two alleles are inherited from a common ancestor. A higher value indicates increased homozygosity in offspring. This increases risk of autosomal recessive disorders. It is important in populations practicing consanguineous marriages. Genetic counseling can reduce such risks. This concept is important in population genetics studies.

[Ref: Falconer DS, Mackay TFC. Introduction to Quantitative Genetics. 4th ed. Harlow: Pearson Education; 1996. pp. 75-89.]

16. Ans: B. National Health Mission

RBSK is implemented under the National Health Mission to improve child health outcomes. It provides screening services for children from birth to 18 years. The program focuses on birth defects, diseases, deficiencies and developmental delays. Early detection allows timely treatment. Referral services are provided through designated health facilities. This program contributes to reduction in childhood morbidity.

[Ref: Ministry of Health and Family Welfare. National Health Mission: Framework for Implementation. New Delhi: Government of India; 2013.]

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17. Ans: D. All of the above

Mutations are permanent alterations in DNA sequence. They may occur due to physical agents such as radiation, chemical mutagens and biological agents like viruses. Mutations may be spontaneous or induced. Some mutations lead to genetic disorders or cancers. Understanding environmental mutagens helps prevention strategies. Environmental protection measures help reduce exposure.

[Ref: Alberts B, Johnson A, Lewis J, et al. Molecular Biology of the Cell. 7th ed. New York: Garland Science; 2022. pp. 245-260.]

18. Ans: B. Proportion of harmful alleles in population

Genetic load refers to accumulation of deleterious alleles in a population that reduce overall fitness. It reflects the burden of harmful genes. Higher genetic load increases prevalence of inherited disorders. Screening and counseling can help reduce this burden. This concept is important in population genetics. Public health strategies aim to reduce genetic disease burden.

[Ref: Park K. Park's Textbook of Preventive and Social Medicine. 28th ed. Jabalpur: Banarsidas Bhanot Publishers; 2025. pp. 620-622.]

19. Ans: B. Gene-environment interaction

Gene-environment interaction refers to the influence of environmental factors on expression of genetic traits. Individuals with genetic predisposition may develop disease when exposed to risk factors. Examples include diabetes, hypertension and coronary artery disease. Lifestyle modification can reduce risk. This concept is important in prevention of noncommunicable diseases. Public health programs focus on controlling modifiable risk factors.

[Ref: Park K. Park's Textbook of Preventive and Social Medicine. 28th ed. Jabalpur: Banarsidas Bhanot Publishers; 2025. pp. 622-624.]

20. Ans: A. Hemoglobinopathies

India has a high prevalence of hemoglobin disorders such as thalassemia and sickle cell disease. Population screening programs focus on these disorders for early detection. Carrier screening helps reduce incidence of affected births. Genetic counseling is an important component. Early detection improves management outcomes. These disorders represent a major public health concern.

[Ref: Indian Council of Medical Research. Guidelines for Prevention and Control of Hemoglobinopathies in India. New Delhi: Indian Council of Medical Research; 2018.]

21. Ans: B. Study inheritance pattern

A pedigree chart is a diagrammatic representation of family relationships used to study inheritance patterns. It helps identify autosomal dominant, autosomal recessive and X-linked disorders. It is useful in genetic counseling and risk prediction. Pedigree analysis helps identify carriers. It is an important tool in medical genetics. This method supports preventive strategies.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 120-130.]

22. Ans: A. Trisomy 21

Down syndrome is most commonly caused by trisomy 21 due to nondisjunction during meiosis. It results in an extra chromosome 21. Clinical features include intellectual disability and characteristic facial features. Congenital heart defects are common. Risk increases with maternal age. Prenatal screening helps early detection.

[Ref: Jameson JL, Fauci AS, Kasper DL, Hauser SL, Longo DL, Loscalzo J. Harrison's Principles of Internal Medicine. 21st ed. New York: McGraw-Hill Education; 2022. pp. 3115-3128.]

23. Ans: B. Proband

The proband is the first affected individual who comes to medical attention in a genetic study. Pedigree analysis usually begins from the proband. It helps trace inheritance patterns. Identification of the proband helps assess family risk. This concept is important in genetic counseling. It helps in disease risk estimation.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 122-124.]

24. Ans: A. 22

Humans have 46 chromosomes arranged in 23 pairs. Out of these, 22 pairs are autosomes and one pair is sex chromosomes. Autosomes carry genes responsible for most body functions. Chromosomal abnormalities involving autosomes may cause genetic disorders. Karyotyping helps identify such abnormalities. This is basic knowledge in human genetics.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 20-25.]

25. Ans: A. 1 in 50

According to Hardy–Weinberg equilibrium, the frequency of an autosomal recessive disease is represented by q^2. If q^2 equals 1/10,000, then q equals 1/100. The carrier frequency is represented by $2pq$, which approximates to $2q$ when p is nearly equal to 1. Therefore, carrier frequency becomes approximately 1/50. This principle is useful for estimating genetic disease burden in populations. It is widely applied in genetic epidemiology and screening programs.

[Ref: Park K. Park's Textbook of Preventive and Social Medicine. 28th ed. Jabalpur: Banarsidas Bhanot Publishers; 2025. pp. 624-626.]

26. Ans: C. 15–18 weeks

Amniocentesis is a prenatal diagnostic procedure performed between 15–18 weeks of gestation. It involves aspiration of amniotic fluid containing fetal cells. These cells are analyzed for chromosomal abnormalities and genetic disorders. It is usually advised when screening tests show high risk. The

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procedure carries a small risk of miscarriage. Proper genetic counseling is essential before performing this test.

[Ref: Cunningham FG, Leveno KJ, Bloom SL, Spong CY, Dashe JS, Hoffman BL, et al. Williams Obstetrics. 26th ed. New York: McGraw-Hill Education; 2022. pp. 287-295.]

27. Ans: B. Genetic disorders

Premarital screening helps identify carriers of genetic disorders such as thalassemia and sickle cell disease. Identification of carriers allows informed marital decisions. Genetic counseling helps couples understand reproductive risks. Such programs are especially important in high prevalence populations. Early identification helps reduce incidence of affected births. This represents an important preventive approach in community genetics.

[Ref: Park K. Park's Textbook of Preventive and Social Medicine. 28th ed. Jabalpur: Banarsidas Bhanot Publishers; 2025. pp. 626-628.]

28. Ans: B. Law of segregation

Mendel's law of segregation states that allele pairs separate during formation of gametes. Each gamete carries only one allele of a gene. During fertilization the alleles recombine. This explains inheritance of traits across generations. This law forms the basis of classical genetics. It remains fundamental to understanding genetic inheritance.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 30-33.]

29. Ans: B. Mendel

Gregor Mendel is known as the father of genetics due to his experiments on pea plants. He described the basic laws of inheritance. His work established the principles of heredity. His research was rediscovered in 1900. Mendelian principles remain fundamental to modern genetics. His work forms the foundation of genetic science.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 28-32.]

30. Ans: B. Management of rare genetic disorders

The National Policy for Rare Diseases aims to improve diagnosis and management of rare disorders. Many rare diseases have genetic origin. The policy supports establishment of centres of excellence. It also provides financial assistance for treatment. Awareness and research are encouraged. This improves access to care for affected patients.

[Ref: Ministry of Health and Family Welfare. National Policy for Rare Diseases. New Delhi: Government of India; 2021.]

31. Ans: B. Maternal age

Advanced maternal age is the most important risk factor for Down syndrome. Increasing maternal age increases the risk of meiotic nondisjunction. Women above 35 years have higher probability of having affected offspring. Prenatal screening is recommended in such pregnancies. Early diagnosis allows proper counseling. This is an important consideration in antenatal genetics.

[Ref: Cunningham FG, Leveno KJ, Bloom SL, Spong CY, Dashe JS, Hoffman BL, et al. Williams Obstetrics. 26th ed. New York: McGraw-Hill Education; 2022. pp. 300-305.]

32. Ans: B. Deletion of chromosome 5p

Cri-du-chat syndrome results from deletion of the short arm of chromosome 5. Affected infants have a characteristic cat-like cry. Other features include microcephaly and intellectual disability. Diagnosis is confirmed by chromosomal analysis. Early supportive therapy improves outcomes. This disorder represents structural chromosomal abnormality.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 92-95.]

33. Ans: B. Stable allele frequency

Hardy–Weinberg equilibrium states that allele frequencies remain constant if no evolutionary forces act. Conditions include large population and random mating. Absence of mutation, migration and selection is required. It is useful in calculating carrier frequency. This principle is widely used in population genetics. It helps plan screening programs.

[Ref: Park K. Park's Textbook of Preventive and Social Medicine. 28th ed. Jabalpur: Banarsidas Bhanot Publishers; 2025. pp. 624-626.]

34. Ans: B. 47 XXY

Klinefelter syndrome is caused by presence of an extra X chromosome in males. Patients may present with infertility and gynecomastia. Testosterone deficiency is common. Diagnosis is confirmed by karyotyping. Hormone replacement therapy may be needed. Early diagnosis improves clinical outcomes.

[Ref: Jameson JL, Fauci AS, Kasper DL, Hauser SL, Longo DL, Loscalzo J. Harrison's Principles of Internal Medicine. 21st ed. New York: McGraw-Hill Education; 2022. pp. 3135-3138.]

35. Ans: B. Double marker test

Double marker test is performed in the first trimester to assess risk of chromosomal abnormalities. It measures PAPP-A and beta-hCG levels. It is often combined with nuchal translucency measurement. High-risk results require confirmatory testing. Early screening improves pregnancy management. It is an important prenatal screening tool.

[Ref: Cunningham FG, Leveno KJ, Bloom SL, Spong CY, Dashe JS, Hoffman BL, et al. Williams Obstetrics. 26th ed. New York: McGraw-Hill Education; 2022. pp. 290-298.]

36. Ans: B. Point mutation

A point mutation involves alteration of a single nucleotide in the DNA sequence. This may occur as substitution, insertion or deletion. Such mutations may alter protein function.

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Sickle cell anemia is a classic example caused by a point mutation. Molecular techniques help detect such changes. Understanding mutation types helps genetic diagnosis.

[Ref: Alberts B, Johnson A, Lewis J, Morgan D, Raff M, Roberts K, et al. Molecular Biology of the Cell. 7th ed. New York: Garland Science; 2022. pp. 255-265.]

37. Ans: B. Wilson and Jungner criteria

Wilson and Jungner proposed screening criteria for disease detection programs. One major criterion states that the disease should be an important public health problem. Other criteria include availability of treatment and suitable screening tests. These principles guide genetic screening programs. They ensure cost-effective screening. These criteria remain fundamental in preventive medicine.

[Ref: Wilson JMG, Jungner G. Principles and Practice of Screening for Disease. Geneva: World Health Organization; 1968.]

38. Ans: B. Trisomy 18

Edwards syndrome is caused by trisomy of chromosome 18. Affected infants have severe developmental abnormalities. Common features include clenched fists and congenital heart defects. Prognosis is generally poor. Diagnosis is confirmed by chromosomal analysis. Prenatal screening helps early detection and counseling.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 95-98.]

39. Ans: B. Founder effect

Founder effect occurs when a new population is established by few individuals. This may increase frequency of certain genetic disorders. It is a type of genetic drift. It is commonly seen in isolated populations. This concept is important in genetic epidemiology. Screening programs often target such populations.

[Ref: Park K. Park's Textbook of Preventive and Social Medicine. 28th ed. Jabalpur: Banarsidas Bhanot Publishers; 2025. pp. 619-620.]

40. Ans: A. Trisomy 13

Patau syndrome results from trisomy of chromosome 13. It is associated with severe congenital anomalies. Common findings include cleft lip, microcephaly and heart defects. Prognosis is usually poor. Diagnosis is confirmed by karyotyping. Prenatal screening helps early identification.

[Ref: Jameson JL, Fauci AS, Kasper DL, Hauser SL, Longo DL, Loscalzo J. Harrison's Principles of Internal Medicine. 21st ed. New York: McGraw-Hill Education; 2022. pp. 3132-3134.]

41. Ans: B. Missense mutation

A missense mutation results from a nucleotide substitution causing change in one amino acid. This may alter protein function. Some missense mutations cause genetic diseases. Sickle cell anemia is a well-known example. Molecular testing helps detect such mutations. This concept is important in molecular pathology.

[Ref: Alberts B, Johnson A, Lewis J, Morgan D, Raff M, Roberts K, et al. Molecular Biology of the Cell. 7th ed. New York: Garland Science; 2022. pp. 260-270.]

42. Ans: C. Isolated population

Isolated populations often have limited genetic diversity. This increases the effects of genetic drift and founder effect. Certain genetic diseases may become more frequent. Consanguinity may further increase risk. Screening programs are often targeted to such groups. Genetic counseling helps reduce disease burden.

[Ref: Park K. Park's Textbook of Preventive and Social Medicine. 28th ed. Jabalpur: Banarsidas Bhanot Publishers; 2025. pp. 618-622.]

43. Ans: B. Amniocentesis

Amniocentesis is a diagnostic prenatal test used to detect chromosomal disorders. It involves analysis of fetal cells obtained from amniotic fluid. It is usually done between 15–18 weeks. It is indicated in high-risk pregnancies. Proper counseling is necessary before testing. Early diagnosis helps informed decision-making.

[Ref: Cunningham FG, Leveno KJ, Bloom SL, Spong CY, Dashe JS, Hoffman BL, et al. Williams Obstetrics. 26th ed. New York: McGraw-Hill Education; 2022. pp. 287-295.]

44. Ans: B. mRNA

Messenger RNA carries genetic information from DNA to ribosomes. It is produced during transcription. The mRNA sequence determines protein structure. This process is essential for gene expression. Errors in transcription may lead to abnormal proteins. This is a key concept in molecular genetics.

[Ref: Alberts B, Johnson A, Lewis J, Morgan D, Raff M, Roberts K, et al. Molecular Biology of the Cell. 7th ed. New York: Garland Science; 2022. pp. 300-310.]

45. Ans: B. PCPNDT Act

The PCPNDT Act regulates the use of prenatal diagnostic techniques. It aims to prevent sex selection and female feticide. It mandates registration of genetic clinics. Violations attract strict legal penalties. The Act promotes ethical use of genetic testing. It is important in reproductive health governance.

[Ref: Ministry of Health and Family Welfare. The Pre-Conception and Pre-Natal Diagnostic Techniques (Prohibition of Sex Selection) Act. New Delhi: Government of India; 2003 (amended 2011).]

46. Ans: B. Homozygosity

Homozygosity refers to the presence of two identical alleles at a gene locus. This increases the likelihood of expression of recessive traits. Consanguineous marriages increase homozygosity in offspring. This may increase

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risk of autosomal recessive disorders. This concept is important in genetic counseling. It helps risk assessment in families.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 40-45.]

47. Ans: C. 18 years

Rashtriya Bal Swasthya Karyakram covers children from birth to 18 years. It includes newborns, preschool and school children. The program focuses on birth defects, diseases, deficiencies and developmental delays. Early identification allows timely intervention. Referral services are provided through designated centers. This improves child health outcomes.

[Ref: Ministry of Health and Family Welfare. Rashtriya Bal Swasthya Karyakram: Operational Guidelines. New Delhi: Government of India; 2013. pp. 10-25.]

48. Ans: B. Replacement of defective gene

Gene therapy involves replacing or modifying defective genes to treat disease. It is used in some inherited disorders. Viral vectors are commonly used for gene delivery. This field is rapidly advancing in molecular medicine. Ethical issues must be considered. It represents a future therapeutic approach.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 410-420.]

49. Ans: B. Nucleus

The nucleus contains chromosomes composed of DNA. DNA carries genetic instructions for protein synthesis. Mutations in nuclear DNA may cause genetic diseases. Some genetic material is also present in mitochondria. Nuclear DNA carries most hereditary information. This is a basic concept in cell biology.

[Ref: Alberts B, Johnson A, Lewis J, Morgan D, Raff M, Roberts K, et al. Molecular Biology of the Cell. 7th ed. New York: Garland Science; 2022. pp. 120-135.]

50. Ans: C. Multifactorial inheritance

Multifactorial inheritance involves interaction of multiple genes and environmental factors. Diseases like diabetes and hypertension follow this pattern. Genetic susceptibility combined with lifestyle factors increases risk. Prevention focuses on lifestyle modification. This concept is important in NCD prevention. Public health programs target modifiable risk factors.

[Ref: Park K. Park's Textbook of Preventive and Social Medicine. 28th ed. Jabalpur: Banarsidas Bhanot Publishers; 2025. pp. 622-624.]

51. Ans: C. X-linked recessive

Duchenne muscular dystrophy is caused by mutation of the dystrophin gene. It follows X-linked recessive inheritance. Males are commonly affected while females are carriers. Progressive muscle weakness starts in early childhood. Genetic testing confirms diagnosis. Carrier detection helps prevent recurrence.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 210-218.]

52. Ans: C. Thymine

DNA contains adenine, guanine, cytosine and thymine. RNA contains uracil instead of thymine. Thymine pairs with adenine through hydrogen bonds. This helps maintain DNA stability. This difference distinguishes DNA from RNA. This is a fundamental molecular biology concept.

[Ref: Alberts B, Johnson A, Lewis J, Morgan D, Raff M, Roberts K, et al. Molecular Biology of the Cell. 7th ed. New York: Garland Science; 2022. pp. 245-250.]

53. Ans: A. Down syndrome

Increased nuchal translucency is a marker of chromosomal abnormalities. Down syndrome is the most common association. It is measured during first trimester ultrasound. High-risk cases require confirmatory testing. Early diagnosis allows counseling. It is an important prenatal screening indicator.

[Ref: Cunningham FG, Leveno KJ, Bloom SL, Spong CY, Dashe JS, Hoffman BL, et al. Williams Obstetrics. 26th ed. New York: McGraw-Hill Education; 2022. pp. 292-300.]

54. Ans: B. Phenotype

Phenotype refers to observable traits of an individual. It results from interaction between genotype and environment. Examples include height and disease expression. Phenotypic variation may occur despite similar genotypes. It helps clinical diagnosis. This is a basic concept in genetics.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 35-40.]

55. Ans: B. Hemoglobin electrophoresis

Hemoglobin electrophoresis detects abnormal hemoglobin variants. β-thalassemia carriers usually show elevated HbA_2 levels. This test is widely used in screening programs. Early detection allows genetic counseling. Carrier screening helps prevent affected births. It is an important preventive strategy.

[Ref: Bain BJ, Bates I, Laffan MA. Dacie and Lewis Practical Haematology. 13th ed. Philadelphia: Elsevier; 2021. pp. 300-315.]

56. Ans: B. Meiosis

Meiosis produces haploid gametes required for sexual reproduction. It reduces chromosome number by half. Crossing over during meiosis increases genetic variation. Errors in meiosis may cause chromosomal disorders. This

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process maintains chromosome number across generations. It is essential in genetics.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 25-30.]

57. Ans: C. Law of independent assortment

This law states that genes for different traits segregate independently. It applies to genes on different chromosomes. It explains variation in offspring. This law is fundamental in classical genetics. It explains inheritance of multiple traits. It remains important in genetic studies.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 32-36.]

58. Ans: C. rRNA

Ribosomal RNA forms the structural framework of ribosomes. It plays a role in protein synthesis. Ribosomes translate mRNA into proteins. rRNA also has catalytic functions. Proper functioning is essential for cell survival. This is a key molecular biology concept.

[Ref: Alberts B, Johnson A, Lewis J, Morgan D, Raff M, Roberts K, et al. Molecular Biology of the Cell. 7th ed. New York: Garland Science; 2022. pp. 310-320.]

59. Ans: B. Random mating

Random mating is essential for Hardy–Weinberg equilibrium. Other assumptions include large population and absence of mutation. Migration and selection should be absent. Violation changes allele frequency. This principle is used in genetic epidemiology. It helps estimate carrier frequency.

[Ref: Park K. Park's Textbook of Preventive and Social Medicine. 28th ed. Jabalpur: Banarsidas Bhanot Publishers; 2025. pp. 624-626.]

60. Ans: B. RBSK

RBSK focuses on early identification of birth defects. Genetic disorders are included under birth defects. Screening is done from birth to 18 years. Early treatment reduces disability. Referral services are available. It is part of NHM child health services.

[Ref: Ministry of Health and Family Welfare. Rashtriya Bal Swasthya Karyakram: Operational Guidelines. New Delhi: Government of India; 2013.]

61. Ans: B. Maternal inheritance

Mitochondrial DNA is inherited only from the mother. This occurs because mitochondria in sperm are destroyed after fertilization. Both males and females can be affected. However, only females transmit the disorder. This inheritance pattern helps in pedigree analysis. It is important in genetic counseling.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 102-108.]

62. Ans: B. Alleles

Alleles are different forms of the same gene present at the same locus. Individuals inherit one allele from each parent. Alleles determine genotype. Variation in alleles produces phenotypic differences. This is a fundamental concept in genetics. It helps understand inheritance patterns.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 10-15.]

63. Ans: C. Karyotyping

Recurrent spontaneous abortions may occur due to chromosomal abnormalities such as balanced translocations in one of the parents. Karyotyping helps identify numerical and structural chromosomal defects. It is recommended when couples have repeated pregnancy losses. Identification of abnormalities allows appropriate genetic counseling. Assisted reproductive options may be considered. This investigation plays an important role in reproductive genetics.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 150-158.]

64. Ans: B. Down syndrome

Triple marker test measures AFP, hCG and estriol. It is performed in the second trimester. It helps assess risk of chromosomal abnormalities. Down syndrome is commonly screened. High-risk cases require confirmatory tests. This improves prenatal diagnosis.

[Ref: Cunningham FG, Leveno KJ, Bloom SL, Spong CY, Dashe JS, Hoffman BL, et al. Williams Obstetrics. 26th ed. New York: McGraw-Hill Education; 2022. pp. 298-305.]

65. Ans: B. DNA

Genes are segments of DNA that contain instructions for protein synthesis. DNA carries hereditary information. Mutations in DNA may cause genetic diseases. DNA directs cellular function. Understanding DNA structure helps genetic diagnosis. This is a basic molecular genetics concept.

[Ref: Alberts B, Johnson A, Lewis J, Morgan D, Raff M, Roberts K, et al. Molecular Biology of the Cell. 7th ed. New York: Garland Science; 2022. pp. 120-135.]

66. Ans: C. Gene–environment interaction

Gene–environment interaction refers to combined effect of genes and environmental factors. Individuals with genetic susceptibility may develop disease after exposure to risk factors. Examples include diabetes and hypertension. Lifestyle modification can reduce risk. This concept is important in NCD prevention. Public health programs focus on risk reduction.

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[Ref: Park K. Park's Textbook of Preventive and Social Medicine. 28th ed. Jabalpur: Banarsidas Bhanot Publishers; 2025. pp. 622-624.]

67. Ans: B. Pea plant

Gregor Mendel used pea plants to study inheritance. He selected traits with clear differences. His experiments led to discovery of genetic laws. These include segregation and independent assortment. His work forms the basis of genetics. He is regarded as father of genetics.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 28-32.]

68. Ans: A. Pedigree analysis

Pedigree analysis helps trace inheritance patterns in families. It helps identify carriers. It is useful in genetic counseling. Risk prediction becomes possible. It is a simple and cost-effective method. It supports preventive genetics.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 120-125.]

69. Ans: B. Genotype

Genotype refers to the genetic makeup of an individual. It consists of alleles inherited from parents. Genotype influences phenotype. Environmental factors may modify expression. Genetic testing identifies genotype. This is a fundamental genetics concept.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 34-38.]

70. Ans: A. Heel prick test

Heel prick test is used in newborn screening programs. Blood collected is tested for metabolic disorders. Early detection allows treatment. This prevents disability and mortality. Such screening is part of preventive pediatrics. It is an important public health intervention.

[Ref: World Health Organization. Newborn and Infant Screening: Current Issues and Guiding Principles. Geneva: World Health Organization; 2010.]

71. Ans: B. DNA polymerase

DNA polymerase is the main enzyme involved in DNA replication. It adds nucleotides to the growing DNA strand. Proofreading ensures accuracy. Errors may cause mutations. DNA replication is essential for cell division. This is a basic concept in molecular genetics.

[Ref: Alberts B, Johnson A, Lewis J, Morgan D, Raff M, Roberts K, et al. Molecular Biology of the Cell. 7th ed. New York: Garland Science; 2022. pp. 270-285.]

72. Ans: C. Diabetes

Multifactorial diseases involve multiple genes and environmental factors. Diabetes is influenced by genetic susceptibility and lifestyle. Obesity increases risk. Prevention focuses on diet and exercise. This concept is important in NCD control. Public health programs target such diseases.

[Ref: Park K. Park's Textbook of Preventive and Social Medicine. 28th ed. Jabalpur: Banarsidas Bhanot Publishers; 2025. pp. 622-624.]

73. Ans: B. Early detection

Screening programs aim to detect disease early. Early diagnosis allows timely intervention. This reduces complications. Screening improves prognosis. Population screening improves health outcomes. It is a key preventive strategy.

[Ref: Wilson JMG, Jungner G. Principles and Practice of Screening for Disease. Geneva: World Health Organization; 1968.]

74. Ans: C. Epistasis

Epistasis refers to interaction between genes. One gene may mask the effect of another. This explains complex inheritance patterns. It is important in polygenic traits. It explains phenotypic variation. It is relevant in molecular genetics.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 60-65.]

75. Ans: B. Y chromosome

Presence of Y chromosome determines male sex. The SRY gene initiates male development. Absence results in female phenotype. This is basis of sex determination. Chromosomal abnormalities may affect development. This is a basic genetics concept.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 70-75.]

76. Ans: B. Mass screening

Mass screening involves testing large populations. It helps detect disease early. It is used for genetic disorders in high-risk areas. Early diagnosis improves management. Screening must follow Wilson–Jungner criteria. It is important in public health.

[Ref: Park K. Park's Textbook of Preventive and Social Medicine. 28th ed. Jabalpur: Banarsidas Bhanot Publishers; 2025. pp. 130-135.]

77. Ans: C. Transcription

Transcription is process of RNA synthesis from DNA. RNA polymerase catalyzes this process. It is first step of gene expression. Errors may affect protein synthesis. This process is essential for cellular function. It is a key molecular biology concept.

[Ref: Alberts B, Johnson A, Lewis J, Morgan D, Raff M, Roberts K, et al. Molecular Biology of the Cell. 7th ed. New York: Garland Science; 2022. pp. 300-305.]

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78. Ans: B. Variable expressivity

Variable expressivity refers to variation in disease severity among individuals. Individuals with same genotype may show different symptoms. Environmental and genetic factors influence expression. It is important in genetic counseling. It explains clinical variability. This is a key genetics concept.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 55-60.]

79. Ans: B. Mitochondrial disorders

Mitochondrial disorders are inherited from mother. This is due to cytoplasmic inheritance. Both sexes may be affected. Only females transmit disease. This helps pedigree analysis. It is important in genetic counseling.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 102-108.]

80. Ans: B. Developmental delay

Developmental delays include delay in speech, cognition, behavior and motor development. RBSK includes screening for developmental delays under the 4Ds. Early identification allows referral to District Early Intervention Centres. Early stimulation therapy improves developmental outcomes. Timely intervention reduces disability burden. This reflects the preventive approach of child health programs.

[Ref: Ministry of Health and Family Welfare. Rashtriya Bal Swasthya Karyakram: Operational Guidelines. New Delhi: Government of India; 2013. pp. 18-26.]

81. Ans: B. Founder effect

Founder effect occurs when a new population is established by a small number of individuals. Due to limited genetic diversity, certain rare alleles may become more frequent. This increases prevalence of specific genetic disorders. It is commonly seen in isolated communities. Genetic screening helps identify such high-risk populations. This concept is important in genetic epidemiology.

[Ref: Park K. Park's Textbook of Preventive and Social Medicine. 28th ed. Jabalpur: Banarsidas Bhanot Publishers; 2025. pp. 619-621.]

82. Ans: B. Turner syndrome

Turner syndrome occurs due to absence of one X chromosome. Patients present with short stature and gonadal dysgenesis. Primary amenorrhea is common. Diagnosis is confirmed by karyotyping. Hormone therapy may be required. Early diagnosis improves management.

[Ref: Jameson JL, Fauci AS, Kasper DL, Hauser SL, Longo DL, Loscalzo J. Harrison's Principles of Internal Medicine. 21st ed. New York: McGraw-Hill Education; 2022. pp. 3128-3132.]

83. Ans: B. Penetrance

Penetrance refers to proportion of individuals with genotype showing phenotype. Complete penetrance means all individuals express the trait. Incomplete penetrance means some do not show expression. This is important in genetic counseling. It explains disease occurrence patterns. It is a key clinical genetics concept.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 52-55.]

84. Ans: B. Karyotyping

Klinefelter syndrome is caused by presence of an extra X chromosome. Patients commonly present with infertility and hypogonadism. Gynecomastia and reduced secondary sexual characters may be present. Diagnosis is confirmed by karyotyping showing 47 XXY. Hormonal therapy may be required. Early diagnosis helps proper management and counseling.

[Ref: Jameson JL, Fauci AS, Kasper DL, Hauser SL, Longo DL, Loscalzo J. Harrison's Principles of Internal Medicine. 21st ed. New York: McGraw-Hill Education; 2022. pp. 3135-3138.]

85. Ans: B. Epigenetics

Epigenetics refers to heritable changes in gene expression without DNA sequence change. Mechanisms include DNA methylation and histone modification. Environmental factors may influence these changes. Epigenetic changes may affect disease risk. This concept explains gene regulation. It is an emerging area in genetics.

[Ref: Alberts B, Johnson A, Lewis J, Morgan D, Raff M, Roberts K, et al. Molecular Biology of the Cell. 7th ed. New York: Garland Science; 2022. pp. 405-415.]

86. Ans: A. Hemophilia

Hemophilia is caused by deficiency of clotting factors VIII or IX. It follows X-linked recessive inheritance. Males are commonly affected. Females are usually carriers. Family history helps diagnosis. Genetic counseling helps prevent recurrence.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 198-205.]

87. Ans: B. Amniocentesis

Screening tests only identify risk and are not diagnostic. Amniocentesis is used to confirm chromosomal abnormalities. It involves analysis of fetal cells from amniotic fluid. It is usually performed between 15–18 weeks. Proper counseling is necessary before testing. Early diagnosis helps pregnancy management decisions.

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[Ref: Cunningham FG, Leveno KJ, Bloom SL, Spong CY, Dashe JS, Hoffman BL, et al. Williams Obstetrics. 26th ed. New York: McGraw-Hill Education; 2022. pp. 287-295.]

88. Ans: B. Prevention of affected births

Carrier detection helps identify couples at risk of having affected children. Genetic counseling allows informed reproductive decisions. Prenatal diagnosis may be offered in high-risk pregnancies. Prevention through screening is more effective than treatment. This reduces long-term healthcare burden. Such programs are important in high prevalence regions.

[Ref: Indian Council of Medical Research. Guidelines for Prevention and Control of Hemoglobinopathies in India. New Delhi: Indian Council of Medical Research; 2018. pp. 22-35.]

89. Ans: C. tRNA

Transfer RNA transports amino acids to ribosomes. It recognizes codons through anticodons. This ensures correct protein synthesis. It plays essential role in translation. Defects may affect protein formation. This is a key molecular biology concept.

[Ref: Alberts B, Johnson A, Lewis J, Morgan D, Raff M, Roberts K, et al. Molecular Biology of the Cell. 7th ed. New York: Garland Science; 2022. pp. 315-320.]

90. Ans: B. Early detection

Newborn screening aims to detect genetic disorders early. Early treatment prevents complications. Screening focuses on treatable conditions. It reduces morbidity and mortality. Public health programs promote such screening. It is an important preventive strategy.

[Ref: World Health Organization. Newborn and Infant Screening: Current Issues and Guiding Principles. Geneva: World Health Organization; 2010.]

91. Ans: B. Bottleneck effect

Bottleneck effect occurs when a population undergoes sudden reduction in size. This leads to loss of genetic variation. Allele frequencies may change due to chance. Some rare alleles may become common. This may increase genetic disease prevalence. It is an example of genetic drift.

[Ref: Park K. Park's Textbook of Preventive and Social Medicine. 28th ed. Jabalpur: Banarsidas Bhanot Publishers; 2025. pp. 619-621.]

92. Ans: B. Sickle cell disease

Sickle cell disease is caused by point mutation in β-globin gene. This produces abnormal hemoglobin. Red blood cells become sickle shaped. This causes anemia and vaso-occlusive crisis. It follows autosomal recessive inheritance. Carrier screening helps prevention.

[Ref: Bain BJ, Bates I, Laffan MA. Dacie and Lewis Practical Haematology. 13th ed. Philadelphia: Elsevier; 2021. pp. 310-320.]

93. Ans: B. Cascade screening

Cascade screening involves testing relatives of affected individuals. It helps identify carriers. It is a cost-effective screening strategy. It allows targeted counseling. It is commonly used in genetic disorders. It helps prevent recurrence.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 125-130.]

94. Ans: C. Frameshift mutation

Frameshift mutation occurs due to insertion or deletion of nucleotides. This alters the reading frame. It often produces abnormal proteins. Such mutations may cause severe disease. Molecular testing helps detection. This concept is important in genetic pathology.

[Ref: Alberts B, Johnson A, Lewis J, Morgan D, Raff M, Roberts K, et al. Molecular Biology of the Cell. 7th ed. New York: Garland Science; 2022. pp. 260-270.]

95. Ans: A. Turner syndrome

Turner syndrome occurs due to monosomy X. Patients show short stature and ovarian failure. Primary amenorrhea is common. Diagnosis is confirmed by karyotyping. Hormone therapy improves outcomes. Early diagnosis is important.

[Ref: Jameson JL, Fauci AS, Kasper DL, Hauser SL, Longo DL, Loscalzo J. Harrison's Principles of Internal Medicine. 21st ed. New York: McGraw-Hill Education; 2022. pp. 3128-3132.]

96. Ans: B. Law of segregation

Law of segregation states that alleles separate during gamete formation. Each gamete receives one allele. Fertilization restores allele pair. This explains inheritance patterns. It is fundamental to Mendelian genetics. It remains important in modern genetics.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 30-33.]

97. Ans: B. Human Genome Project

Human Genome Project aimed to sequence entire human genome. It was completed in 2003. It improved understanding of genetic diseases. It enabled genetic diagnostics. It advanced personalized medicine. It remains a milestone in genetics.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 15-20.]

98. Ans: B. Linkage analysis

Linkage analysis studies genes located close together. Such genes tend to be inherited together. It helps identify disease

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genes. It is used in molecular genetics. It supports gene mapping. It has diagnostic importance.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 140-145.]

99. Ans: B. XX

Females have two X chromosomes. One X chromosome is inherited from each parent. Absence of Y chromosome results in female development. Sex determination depends on Y chromosome presence. Chromosomal abnormalities may affect development. This is basic human genetics.

[Ref: Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 9th ed. Philadelphia: Elsevier; 2023. pp. 70-75.]

100. Ans: B. Confidentiality

Genetic information is sensitive personal data. Confidentiality must be maintained during testing. Disclosure without consent may cause discrimination. Ethical guidelines emphasize privacy. Proper counseling is essential. This is important in medical ethics.

[Ref: World Health Organization. Genomics and World Health: Report of the Advisory Committee on Health Research. Geneva: World Health Organization; 2002.]

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